



PORCN gene

porcupine homolog (*Drosophila*)

Normal Function

The *PORCN* gene provides instructions for making a protein that belongs to a group of related proteins called the porcupine (Porc) family. Although the precise function of the *PORCN* protein is unknown, proteins in the Porc family are involved in the process of transferring a molecule called palmitoleic acid to Wnt proteins. Wnt proteins participate in chemical signaling pathways in the body and play critical roles in development before birth. Members of the Porc family are located in the endoplasmic reticulum, which is a structure inside the cell that is involved in protein processing and transport. The transfer of palmitoleic acid to Wnt proteins facilitates the release of these proteins from the cell so they can regulate development of the skin, bones, and other structures. Researchers are working to determine the specific role of the *PORCN* protein within human cells.

Health Conditions Related to Genetic Changes

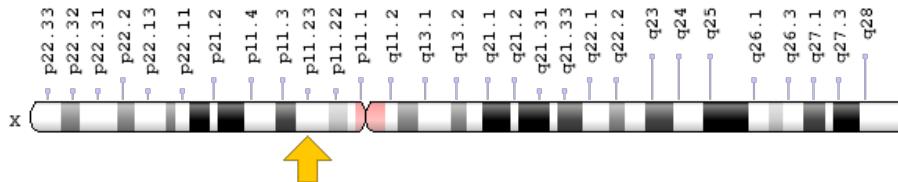
focal dermal hypoplasia

At least 29 mutations in the *PORCN* gene have been found to cause focal dermal hypoplasia. These mutations may alter the protein's structure, lead to the production of an abnormally short version of the protein, or delete the entire *PORCN* gene. All of these mutations appear to result in the absence of any functional *PORCN* protein. Researchers believe Wnt proteins cannot be released from the cell without the *PORCN* protein. When Wnt proteins are unable to leave the cell, they cannot participate in the chemical signaling pathways that are critical for normal development.

Chromosomal Location

Cytogenetic Location: Xp11.23, which is the short (p) arm of the X chromosome at position 11.23

Molecular Location: base pairs 48,508,954 to 48,520,814 on the X chromosome (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- DHOF
- FODH
- MG61
- MGC29687
- por
- PORC
- PORCN_HUMAN
- porcupine
- porcupine isoform A
- porcupine isoform B
- porcupine isoform C
- porcupine isoform D
- porcupine isoform E
- PPN

Additional Information & Resources

Educational Resources

- The Wnt Homepage, Stanford University
<https://web.stanford.edu/group/nusselab/cgi-bin/wnt/>

GeneReviews

- Focal Dermal Hypoplasia
<https://www.ncbi.nlm.nih.gov/books/NBK1543>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28PORCN%5BTIAB%5D%29+OR+%28PORC%5BTIAB%5D%29+OR+%28porcupine%5BTIAB%5D%29%29+AND+%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+720+days%22%5Bdp%5D>

OMIM

- PORCUPINE, DROSOPHILA, HOMOLOG OF
<http://omim.org/entry/300651>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_PORCN.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=PORCN%5Bgene%5D>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=17652
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/64840>
- UniProt
<http://www.uniprot.org/uniprot/Q9H237>

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